Amniotic Fluid Analysis—A Cooperative And Multidiscipline Approach to the Prevention of Certain Inherited Disorders

By study of the amniotic fluid, it now appears possible to detect over 30 biochemical genetic disorders and a variety of chromosomal aberrations. Fluid usually is collected by amniocentesis sometime between the sixteenth and twentieth week of pregnancy. In experienced hands this procedure carries a very minimal risk for maternal or fetal complications.

The usual indications for such tests are those high-risk pregnant women who have already borne children with one of these specific disorders. The biochemical disorders are mostly rare, often serious, recessively inherited diseases. The most common heritable chromosome problem is the translocation-type Down's syndrome (mongolism).

An unfortunate drawback is the amount of time needed in most cases to do the analysis. For a few biochemical disorders the tests can be done directly on the fresh amniotic fluid specimen. In most biochemical cases, however, and for all chromosome studies the cells must be cultured four or more weeks. Barrbody prenatal sex determination can be done on the fresh specimen, and this sometimes is used to sharpen the risk calculation for certain serious, sex-linked, recessive diseases, for example hemophilia, pseudohypertrophic muscular dystrophy.

The obvious purpose for such prenatal detection is to reassure families when the specific diseases are absent and to alert them to abnormalities in sufficient time so that interruption of pregnancies is possible. Existing legislation in each state will determine the practicality of such diagnostic procedures.

Optimum utilization and progress in this relatively new field of preventive medicine require the combined efforts of competent obstetricians, clinical geneticist-counselors, advanced biochemical and chromosome laboratory facilities, public health support, scientific researchers and enlightened forward-looking legislation.

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REFERENCES

Nadler HL: Prenatal detection of genetic defects. J Pediat 74:132-143, 1969

Littlefield JW, Goldstein S, Shih VE: Cell cultures and pre- and postnatal diagnosis. Pediat News 3:15, 1969

Nadler HL, Gerbie AB: Enzymes in noncultured amniotic fluid cells. Amer J Obstet Gynec 103:710-712, 1969

The Importance of Birthweight To Gestational Age

Over a decade ago, the term *low birth weight* was adopted internationally in preference to *prematurity*. Until then the term *prematurity* had described a mixture, from infants actually born prematurely to those full or even post-term.

True prematures are infants whose development in utero had progressed satisfactorily until the untimely early delivery of the infant. The premature infant, born too soon, is sized appropriately for the length of gestation.

All other infants of low birth weight have had an abnormal intrauterine growth for a variety of possible causes. They tend to be abnormally small for their gestational age. They include infants with chromosomal aberrations (Down's syndrome), genetic diseases (cystic fibrosis), metabolic diseases (osteogenesis imperfecta), maternal illnesses (toxemia), intrauterine infections (rubella) prolonged pregnancy (postmaturity) and congenital malformations (Potter's syndrome).

Newborns also can be excessively large for gestational age, as is seen in offspring of mothers with diabetes.

Mechanisms of smallness vary. Infants, for example, born after an intrautero infection such as rubella, appear to have diminished numbers of cells.

Infants born to mothers with toxemia are examples of true intrauterine growth retardation, with normal numbers of cells but each cell apparently smaller than that of the normally sized infant for that gestational age.

Knowledge of appropriateness of size for gestation can provide important clues toward delineating diseases and also toward suggesting prognosis.

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REFERENCES

Lubchenco LO, Hansman C, Bäckström L: Factors influencing fetal growth, *In* Jonxis JHP, Visser HKA, Troelstra JA (Eds): Nutrition Symposium—Aspects of Praematurity and Dysmaturity. Leiden, H E Stenfert Kroese, Publishers, 1968, pp 149-166

Silverman WA, Sinclair JC: Infants of low birth weight. New Eng J Med 274:448-450, 1966

Naeye RL: Unsuspected organ abnormalities associated with congenital heart disease. Am J Path 47:905-915, 1965